



EFTUD2 gene

elongation factor Tu GTP binding domain containing 2

Normal Function

The *EFTUD2* gene provides instructions for making one part (subunit) of two complexes called the major and minor spliceosomes. Spliceosomes help process messenger RNA (mRNA), which is a chemical cousin of DNA that serves as a genetic blueprint for making proteins. The spliceosomes recognize and then remove regions called introns to help produce mature mRNA molecules.

Health Conditions Related to Genetic Changes

mandibulofacial dysostosis with microcephaly

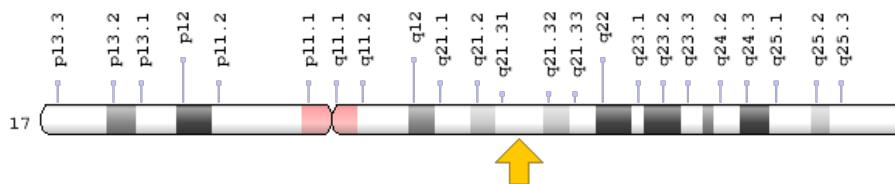
More than 50 *EFTUD2* gene mutations have been identified in people who have mandibulofacial dysostosis with microcephaly (MFDM). This disorder causes malformations of the head and face, intellectual disability, and abnormalities affecting other areas of the body. These abnormalities include esophageal atresia, which is a blockage of the esophagus, and tracheoesophageal fistula, which is an abnormal connection between the esophagus and the trachea that allows fluids from the esophagus to get into the airways and interfere with breathing.

The *EFTUD2* gene mutations that cause MFDM result in the production of little or no functional enzyme from one copy of the gene in each cell. A shortage of this enzyme likely impairs mRNA processing. The relationship between these mutations and the specific symptoms of MFDM is not well understood.

Chromosomal Location

Cytogenetic Location: 17q21.31, which is the long (q) arm of chromosome 17 at position 21.31

Molecular Location: base pairs 44,850,287 to 44,899,625 on chromosome 17 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 116 kDa U5 small nuclear ribonucleoprotein component
- elongation factor Tu GTP-binding domain-containing protein 2
- hSNU114
- MFDGA
- MFDM
- SNRNP116
- Snrp116
- Snu114
- SNU114 homolog
- U5-116KD
- U5 snRNP-specific protein, 116 kDa

Additional Information & Resources

Educational Resources

- Biochemistry: (fifth edition, 2002): RNA Synthesis and Splicing
<https://www.ncbi.nlm.nih.gov/books/NBK21189/>

GeneReviews

- Mandibulofacial Dysostosis with Microcephaly
<https://www.ncbi.nlm.nih.gov/books/NBK214367>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28EFTUD2%5BTIAB%5D%29+OR+%28MFDGM%5BTIAB%5D%29+OR+%28Snu114%5BTIAB%5D%29+OR+%28Snrp116%5BTIAB%5D%29+OR+%28U5-116KD%5BTIAB%5D%29+OR+%28hSNU114%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- ELONGATION FACTOR Tu GTP-BINDING DOMAIN-CONTAINING 2
<http://omim.org/entry/603892>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_EFTUD2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=EFTUD2%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=30858
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/9343>
- UniProt
<http://www.uniprot.org/uniprot/Q15029>

Sources for This Summary

- OMIM: ELONGATION FACTOR Tu GTP-BINDING DOMAIN-CONTAINING 2
<http://omim.org/entry/603892>
- GeneReview: Mandibulofacial Dysostosis with Microcephaly
<https://www.ncbi.nlm.nih.gov/books/NBK214367>
- Gordon CT, Petit F, Oufadem M, Decaestecker C, Jourdain AS, Andrieux J, Malan V, Alessandri JL, Baujat G, Baumann C, Boute-Benejean O, Caumes R, Delobel B, Dieterich K, Gaillard D, Gonzales M, Lacombe D, Escande F, Manouvrier-Hanu S, Marlin S, Mathieu-Dramard M, Mehta SG, Simonic I, Munnich A, Vekemans M, Porchet N, de Pontual L, Sarnacki S, Attie-Bitach T, Lyonnet S, Holder-Espinasse M, Amiel J. EFTUD2 haploinsufficiency leads to syndromic oesophageal atresia. *J Med Genet.* 2012 Dec;49(12):737-46. doi: 10.1136/jmedgenet-2012-101173.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23188108>

- Guion-Almeida ML, Zechi-Ceide RM, Vendramini S, Tabith Júnior A. A new syndrome with growth and mental retardation, mandibulofacial dysostosis, microcephaly, and cleft palate. *Clin Dysmorphol.* 2006 Jul;15(3):171-4.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/16760738>
- Lehalle D, Gordon CT, Oufadem M, Goudefroye G, Boutaud L, Alessandri JL, Baena N, Baujat G, Baumann C, Boute-Benejean O, Caumes R, Decaestecker C, Gaillard D, Goldenberg A, Gonzales M, Holder-Espinasse M, Jacquemont ML, Lacombe D, Manouvrier-Hanu S, Marlin S, Mathieu-Dramard M, Morin G, Pasquier L, Petit F, Rio M, Smigiel R, Thauvin-Robinet C, Vasiljevic A, Verloes A, Malan V, Munnich A, de Pontual L, Vekemans M, Lyonnet S, Attié-Bitach T, Amiel J. Delineation of EFTUD2 haploinsufficiency-related phenotypes through a series of 36 patients. *Hum Mutat.* 2014 Apr;35(4):478-85. doi: 10.1002/humu.22517. Epub 2014 Mar 5.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/24470203>
- Lines MA, Huang L, Schwartzentruber J, Douglas SL, Lynch DC, Beaulieu C, Guion-Almeida ML, Zechi-Ceide RM, Gener B, Gillessen-Kaesbach G, Nava C, Baujat G, Horn D, Kini U, Caliebe A, Alanay Y, Utine GE, Lev D, Kohlhase J, Grix AW, Lohmann DR, Hehr U, Böhm D; FORGE Canada Consortium, Majewski J, Bulman DE, Wieczorek D, Boycott KM. Haploinsufficiency of a spliceosomal GTPase encoded by EFTUD2 causes mandibulofacial dysostosis with microcephaly. *Am J Hum Genet.* 2012 Feb 10;90(2):369-77. doi: 10.1016/j.ajhg.2011.12.023. Epub 2012 Feb 2.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/22305528>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3276671/>
- Voigt C, Mégarbané A, Neveling K, Czeschik JC, Albrecht B, Callewaert B, von Deimling F, Hehr A, Falkenberg Smeland M, König R, Kuechler A, Marcelis C, Puiu M, Reardon W, Riise Stensland HM, Schweiger B, Steehouwer M, Teller C, Martin M, Rahmann S, Hehr U, Brunner HG, Lüdecke HJ, Wieczorek D. Oto-facial syndrome and esophageal atresia, intellectual disability and zygomatic anomalies - expanding the phenotypes associated with EFTUD2 mutations. *Orphanet J Rare Dis.* 2013 Jul 24;8:110. doi: 10.1186/1750-1172-8-110.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/23879989>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3727992/>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/gene/EFTUD2>

Reviewed: September 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services